



distal myopathy 2

Distal myopathy 2 is a condition characterized by weakness of specific muscles that begins in adulthood. It is a form of muscular dystrophy that specifically involves muscles in the throat, lower legs, and forearms. Muscles farther from the center of the body, like the muscles of the lower legs and forearms, are known as distal muscles.

Muscle weakness in the ankles is usually the first symptom of distal myopathy 2. The weakness can also affect muscles in the hands, wrists, and shoulders. At first, the muscle weakness may be on only one side of the body, but both sides are eventually involved. This muscle weakness can slowly worsen and make actions like walking and lifting the fingers difficult.

Another characteristic feature of distal myopathy 2 is weakness of the vocal cords and throat. This weakness initially causes the voice to sound weak or breathy (hypophonic). Eventually, the voice becomes gurgling, hoarse, and nasal. The weakness can also cause difficulty swallowing (dysphagia).

Frequency

The prevalence of distal myopathy 2 is unknown. At least two families with the condition have been described in the scientific literature.

Genetic Changes

A mutation in the *MATR3* gene has been identified in people with distal myopathy 2. This gene provides instructions for making a protein called matrin 3, which is found in the nucleus of the cell as part of the nuclear matrix. The nuclear matrix is a network of proteins that provides structural support for the nucleus and aids in several important nuclear functions.

The function of the matrin 3 protein is unknown. This protein can attach to (bind) RNA, which is a chemical cousin of DNA. Some studies indicate that matrin 3 binds and stabilizes a type of RNA called messenger RNA (mRNA), which provides the genetic blueprint for proteins. Matrin 3 may also bind certain abnormal RNAs that might lead to nonfunctional or harmful proteins, thereby blocking the formation of such proteins. Other studies suggest that the matrin 3 protein may be involved in cell survival.

The *MATR3* gene mutation identified in people with distal myopathy 2 changes a single protein building block (amino acid) in the matrin 3 protein. The effect of this mutation on the function of the protein is unknown, although one study suggests that the mutation may change the location of the protein in the nucleus. Researchers are working to

determine how this gene mutation leads to the signs and symptoms of distal myopathy 2.

Inheritance Pattern

Distal myopathy 2 is inherited in an autosomal dominant pattern, which means one copy of the altered *MATR3* gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- distal myopathy with vocal cord and pharyngeal signs
- distal myopathy with vocal cord weakness
- matrin 3 distal myopathy
- MPD2
- myopathia distalis type 2
- VCPDM
- vocal cord and pharyngeal weakness with distal myopathy

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Myopathy, distal, 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1853723/>

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Muscular Dystrophy
<https://medlineplus.gov/ency/article/001190.htm>
- National Institute of Neurological Disorders and Stroke: Muscular Dystrophy: Hope Through Research
<https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Hope-Through-Research/Muscular-Dystrophy-Hope-Through-Research>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>

- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Muscular Dystrophy
<https://medlineplus.gov/ency/article/001190.htm>
- Health Topic: Muscle Disorders
<https://medlineplus.gov/muscledisorders.html>
- Health Topic: Muscular Dystrophy
<https://medlineplus.gov/musculardystrophy.html>

Genetic and Rare Diseases Information Center

- Distal myopathy with vocal cord weakness
<https://rarediseases.info.nih.gov/diseases/1887/distal-myopathy-with-vocal-cord-weakness>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Muscular Dystrophy: Hope Through Research
<https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Hope-Through-Research/Muscular-Dystrophy-Hope-Through-Research>

Educational Resources

- Cleveland Clinic: Muscular Dystrophy
<http://my.clevelandclinic.org/health/articles/muscular-dystrophy>
- MalaCards: distal myopathy with vocal cord weakness
http://www.malacards.org/card/distal_myopathy_with_vocal_cord_weakness
- Muscular Dystrophy Association: Facts About Rare Muscular Dystrophies
https://www.mda.org/sites/default/files/publications/Facts_RareMDs_P-214_0.pdf
- Orphanet: Distal myopathy with vocal cord weakness
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=600
- Washington University, St. Louis: Neuromuscular Disease Center: Distal Myopathy with Vocal Cord & Pharyngeal Weakness (MPD2)
<http://neuromuscular.wustl.edu/musdist/distal.html#distvc>

Patient Support and Advocacy Resources

- Muscular Dystrophy Association
<https://www.mda.org/>
- National Organization for Rare Disorders (NORD): Distal Myopathy
<https://rarediseases.org/rare-diseases/distal-myopathy/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22distal+myopathy+2%22+OR+%22Distal+Myopathies%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Distal+Myopathies%5BMAJR%5D%29+OR+%28vcadm%5BALL%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- AMYOTROPHIC LATERAL SCLEROSIS 21
<http://omim.org/entry/606070>

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